Torus Mandibularis: A Genetic Study

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TORUS MANDIBULARIS is a benign exostosis located on the lingual surface of the mandible usually opposite the cuspid and premolar teeth (Fig. 1).

Several theories of origin for mandibular tori have been proposed, the two most prominent being related to masticatory stress (Bernier, 1955; Hrdlička, 1940; Hooten, 1918; Johnson, 1959; Matthews, 1933; Weiman and Sicher, 1947) and heredity (Courturier, 1958; Drennan, 1937; Furst, 1908; Keeler, 1935; Kolas *et al.*, 1953; Krahl, 1949; Lasker, 1947; Moorrees, Osborne, and Wilde, 1952; Suzuki and Sakai, 1960; Witkop, 1961). Johnson (1959) suggested that the correlation between the amount of tooth wear and torus development was an indication of a functional response of the mandible to 1935; Kolas *et al.*, 1953; Krahl, 1949; Lasker, 1947; Moorrees, Osborne, and abnormal masticatory stress. This hypothesis is supported by the observations of Hrdlička (1940) and Hooten (1918) of mandibular tori in Eskimos with severe dental attrition.

However, Moorrees, Osborne, and Wilde (1952), on examination of Aleut Eskimos, found mandibular tori to have a familial background and, furthermore, noted no correlation with tooth wear. In fact, they postulated that at least three independent loci were required for the presence of mandibular tori, a major gene and two modifiers.

Suzuki and Sakai (1960) studied seven pedigrees of mandibular tori and concluded that the trait exhibited the characteristics of an autosomal dominant inheritance pattern.

The present study was undertaken to define more clearly the role which genetic factors play in the formation of mandibular tori.

METHODS AND MATERIALS

Probands were obtained at the United States Veterans Administration Hospital and the University of Minnesota School of Dentistry, Minneapolis,

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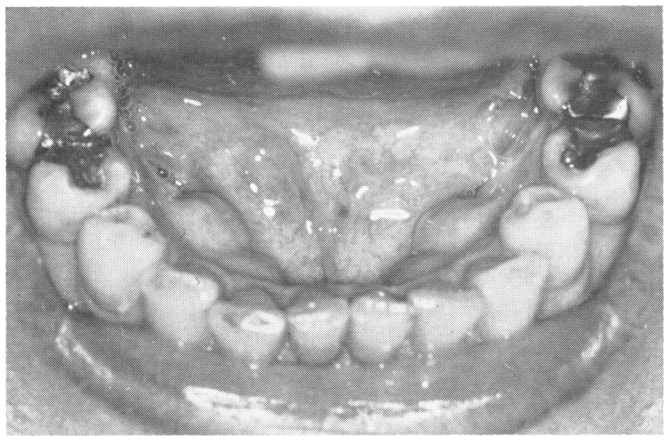

FIG. 1. Multiple bilateral mandibular tori.

Minnesota. All probands had an unequivocal mandibular torus. The relatives were examined at home, and determination of the presence of mandibular tori was made both visually and by palpation. All available relatives were traced, and it was assumed that no bias was introduced into the sample by the failure to follow the relatives not readily examined.

The sex, age, and presence or absence of mandibular tori were recorded on a pedigree chart for each member of the family. (See Appendix.) The pedigree charts were examined for a familial concentration of the trait and for the presence of age and sex factors affecting the clinical appearance of the trait. Individuals who married into the subject families were used as the control group.

RESULTS

The total number of probands in the investigation was 56. Of these, 52 were male, reflecting the use of a Veterans Hospital as a source of probands (Table 1).

The analysis of the first degree relatives of the probands and the children of other affected persons indicated the presence of a sex bias by revealing a frequency of mandibular tori in males of 26.0% and a frequency of 39.0% in females (Table 2). The age of the individuals was found to affect the appearance of mandibular tori, since persons under the age of 15 years had a frequency of mandibular tori of approximately 10.0% (Fig. 2). In individuals over the age of 14 years, the frequency of mandibular tori was 36.0% in males and 52.0% in females (Table 2). This adult group had an over-all frequency of mandibular tori of 44.4% (Table 2).

A test of the difference between the sexes in the frequency rates of mandibular tori indicated that there was less than 1 in 100 chance that the appearance of the trait is not dependent upon the sex of the individual ($p_1 = 0.52$, $N_1 = 125$, $p_2 = 0.36$, and $N_2 = 111$) (Dixon and Massey, 1957, pp. 63, 225).

The males and females of the control group were found to have a 5.9% and

TABLE 1. SOURCE OF PROBANDS

Source	Male	Female	Total
University of Minnesota	5	2	7
Veterans Administration Hospital	47	2	49
Total	52	4	56

TABLE 2. DISTRIBUTION OF RELATIVES OF AFFECTED PERSONS*

Age (years)	Male			Female			Total		
	N	A	%A	N	A	%A	N	A	%A
0–14	60	6	9.1	51	6	10.5	111	12	9.8
15+	71	40	36.0	60	65	52.0	131	105	44.4
Total	131	46	26.0	111	71	39.0	242	117	32.6

Tests of significance (15+ years only):
Male *vs.* Female: $P < 0.01$
Total *vs.* Control (Table 3): $P < 0.00001$
N = Normal, A = Affected, %A = Per cent affected.
*Includes first degree relatives of probands, the children of affected siblings of probands, and the children of affected persons of later generations.

a 8.5% frequency of mandibular tori, respectively. The population frequency of mandibular tori in the group was 7.4% (Table 3).

The analysis for familial concentration was performed by comparing the frequency of mandibular tori in the first degree relatives of an affected person with that found in the control group ($p_1 = 0.444$, $N_1 = 236$, $p_2 = 0.074$, and $N_2 = 122$). This test indicated that there is less than 1 in 100,000 chance that the trait is not familial.

Since a sex factor appeared to be implicated in the appearance of mandibular tori, questions concerning sex-linkage were asked. If the trait were X-linked dominant, then none of the sons of an affected father with a normal wife should have mandibular tori. However, 21.5% of these sons did indeed have mandibular tori, thereby excluding an X-linked dominant trait (Table 4). Conversely, if the trait were X-linked recessive, none of the daughters of an affected mother with a normal husband should have mandibular tori. However, 35.5% of these daughters did in fact have mandibular tori, thereby excluding an X-linked recessive trait (Table 4).

With sex-linkage for the trait eliminated, the autosomal mode of inheritance was investigated. Assuming the trait to be of the autosomal recessive type, all the children of two affected parents should have mandibular tori. However, only 37.5% of the male children and 30.8% of the female children were found to have mandibular tori. Furthermore, when all children under the age of 15 years were excluded, the over-all frequency increased to only 54.5%. This represented one-half the expected rate for mandibular tori, thus rejecting autosomal recessive inheritance with complete penetrance (Table 4).

If the trait were assumed to be autosomal dominant, then approximately 50% of the first degree relatives of the probands and the children of other

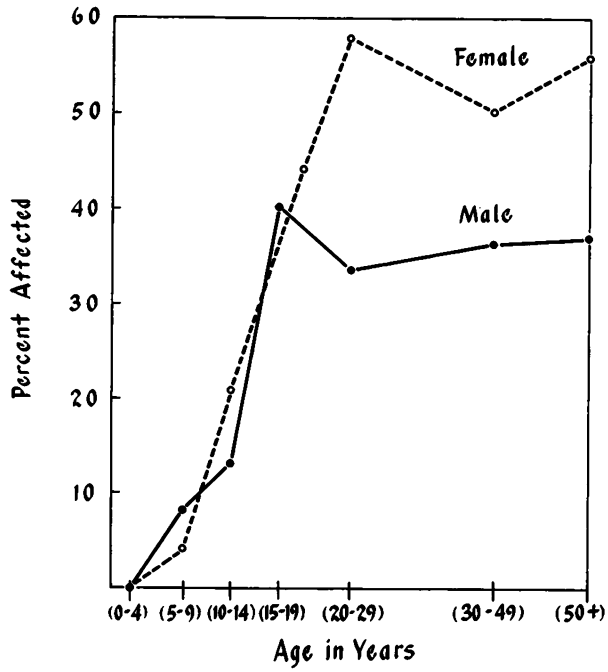

FIG. 2. Distribution of relatives of affected persons. Includes first degree relatives of probands, the children of affected siblings of probands, and the children of affected persons of later generations.

affected persons should have mandibular tori. The actual observed rates of mandibular tori were 26.0% in males and 39.0% in females. However, when all persons under the age of 15 years were excluded, these rates increased to 36.0% in males and 52.0% in females (Table 2).

These rates are compatible with 100% penetrance of mandibular tori in females and a reduced penetrance of mandibular tori in males. An estimation of the penetrance in males over the age of 14 years can be obtained from the ratio of the frequency in males divided by the frequency in females, i.e., 36/52 or 69.2% penetrance in males (Table 2). This is corroborated by the fact that the rate in the control males is 69.4% of the rate in the control females, i.e., 5.9/8.5 or 69.4% (Table 3).

The expected number of affected children with one affected parent was then determined under the assumption of autosomal dominance and the calculated penetrance for the sexes. The expected number of 17.4 affected male children was estimated; 19 were actually observed. The expected number of affected female children was 27.5, and 29 were actually observed (Table 5).

The rate of mandibular tori in the children of two affected parents was estimated as 63.8%. This was obtained from the product of the average penetrance (85.0%) and the expected rate of mandibular tori in the offspring of two heterozygous individuals (75.0%). This investigation yielded seven families

TABLE 3. DISTRIBUTION OF PERSONS MARRYING INTO FAMILIES

Age (years)	Male			Female			Total		
	N	A	%A	N	A	%A	N	A	%A
20+	48	3	5.9	65	6	8.5	113	9	7.4

TABLE 4. DISTRIBUTION OF CHILDREN OF PROBANDS, SIBLINGS, AND LATER GENERATIONS BY MATING TYPES OF THE PARENTS

Parents			Sons			Daughters		
Mother	Father	No.	N	A	%A	N	A	%A
N	N	42	39	6	13.3	58	2	3.3
N	A	44	62	17	21.5	44	19	30.2
A	N	25	23	5	17.9	20	11	35.5
A	A	7	5	3	37.5	9	4	30.8

Children over 14 years of age with A × A parents

N	A	%A
5	6	54.5

TABLE 5. OCCURRENCE OF MANDIBULAR TORI IN CHILDREN WITH ONE AFFECTED PARENT AND ONE NONAFFECTED PARENT

Children under 15 years of age are excluded

	Number	Expected number if autosomal dominant gene	Expected number with 69.4% penetrance in males	Observed
Sons	50	25	17.4	19
Daughters	55	27.5	27.5	29

with two affected parents, and 54.5% (6 of 11) of their children had mandibular tori (Table 4).

It was realized that this number of families and individuals was quite small and therefore possibly inaccurate. For this reason, the study of Suzuki and Sakai (1960) was examined relative to the offspring of two affected parents. It was found that 58.6% of the children of two affected parents had mandibular tori which closely agreed with the expected rate of 60.0%. The expected rate was obtained by the product of their average frequency (80.0%) and the expected rate of mandibular tori in the offspring of two heterozygous individuals (75.0%). Although these percentages are in agreement, there was a possible qualifying condition as these authors made no mention of the age of the subjects. Their data gave further credence to the thought of autosomal dominance for the trait by showing that 4.9% of the children of two normal parents had mandibular tori while 40.0% of the children of one normal and one affected parent had mandibular tori. These percentages are compatible with the expected range of an autosomal dominant gene with incomplete penetrance.

Mandibular tori in children with two normal parents could be the result

of incomplete penetrance of the trait in one of the parents or possibly due to error in judgment in not diagnosing a mandibular torus, when in reality one was present (Table 4). A specific error that could have been operating was that a number of the older persons had undergone full mouth extraction of their teeth with the possible removal of the mandibular torus to facilitate the fitting of dentures. The possibility of a new mutation was considered but was thought to be unlikely.

An estimation of the population gene frequency was made by assuming that the trait was 100% penetrant in females over the age of 14 years in the control group. Then the frequency of 8.5% in females is equal to the $p^2 + 2pq$ of the Hardy-Weinberg equation, the quantity p being equal to 0.043 and q being equal to 0.957. The frequency of homozygous individuals (p^2) would be 0.2% and the frequency of heterozygous individuals ($2pq$) 8.3%.

SUMMARY AND CONCLUSIONS

The origin of mandibular tori appears to be genetic, since this investigation demonstrated a familial concentration of the trait. The frequency of mandibular tori becomes stabilized after the age of 14 years. The pedigrees strongly indicate an autosomal dominant mode of inheritance. An apparent sex predilection exists since males exhibit only 70% of the frequency found in females.

The population gene frequency is approximately 5% and the actual frequency of mandibular tori is 8.5%. The frequency of homozygous persons, 0.2%, is considerably less than the frequency of heterozygous persons, 8.3%. These conclusions are based on the assumption of autosomal dominance for the trait.

However, the appearance of mandibular tori could be the result of multiple genetic factors not detected in this investigation. Also, the size and number of mandibular tori were not recorded, and these could be the result of cumulative actions.

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APPENDIX

Pedigrees of families with one or more persons affected with torus mandibularis. The age at time of investigation is given below each symbol.